

## Neurology

### Prenatal Diagnosis of Inherited Neurologic Diseases

FORTY-TWO HEREDITARY METABOLIC disorders are potentially diagnosable before birth; the diagnosis has been made prenatally in 14 of them. These disorders are transmitted as either autosomal recessive traits or X-linked recessive traits with a 25 percent recurrence risk in subsequent siblings of an affected child. Nearly all these disorders are fatal, untreatable and involve severe mental retardation. In addition to these metabolic disorders, numerous chromosomal defects, most of which increase in frequency with advancing maternal age (highest risk group, mothers over 40), can be diagnosed prenatally. The optimal time for amniocentesis is at 16 to 18 weeks of pregnancy. In California, second trimester diagnostic amniocentesis has been performed most frequently at medical school affiliated hospitals, including UC Davis, UC San Francisco, Stanford, USC, UCLA, Loma Linda, UC Irvine and UC San Diego.

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Milunsky A, Littlefield JW, Kanfer JN, et al: Prenatal genetic diagnosis. *New Eng J Med* 283:1370-1381, Dec 17, 1970  
O'Brien JS: How we detect mental retardation before birth. *Med Times* 99:103-108, Feb 1971

### Brain Death

CARDIAC TRANSPLANTATION AND ITS dramatic life-saving potential require viable donor organs. Salvage of such organs necessitated modification of the traditional concept of death with the introduction of "brain or cerebral death" as tantamount to death despite the presence of cardiac rhythm. Currently agreed upon criteria hinges upon electrical evidences indicating no spontaneous brain activity. Thus, a comatose patient, in the absence of sedative over-dosage or hypothermia, demonstrating two consecutive one-half

hour "flat or isoelectric electroencephalograms" (EEG) over a 24-hour interval is recognized to have irreversible coma.

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- Silverman D, Masland RL, Saunders MG, et al: Irreversible coma associated with electrocerebral silence. *Neurology* 20: 525-533, Jun 1970  
Beecher HK: A definition of irreversible coma—Report of the ad hoc committee of the Harvard Medical School to examine the definition of brain death. *JAMA* 205:337-342, Aug 5, 1968

### Treatment of Cerebral Edema

CEREBRAL EDEMA HAS been demonstrated to be the cause of significant morbidity and mortality in association with trauma, neoplasm, infarction, infection, and toxic and metabolic disorders of the nervous system. Dehydration of the acutely swollen brain can be rapidly produced in 20 to 30 minutes by intravenous infusion of a urea solution by creating an osmotic gradient for water to leave brain. A 20 percent mannitol solution in distilled water can be administered in the same manner and with much the same results. Another technique of reducing cerebral swelling is to passively hyperventilate which will reduce  $\text{PaCO}_2$  and result in cerebral vasoconstriction. Subacute or chronic cerebral edema can be reduced for weeks or months with dexamethasone 4 mg every 6 hours. Within 24 hours, patients may show an improvement in the state of consciousness and in the levels of their vital signs.

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- Matson DD: Treatment of cerebral swelling. *New Eng J Med* 272: 626-628, Mar 25, 1965  
Reulen HJ, Samii M, Fenske K, et al: Electrolytes fluid and energy metabolism in traumatic brain edema. *J Neural Neurosurg Psychiatry* 33:714-715, Oct 1970

### Normal Pressure Hydrocephalus

"NORMAL PRESSURE OR OCCULT hydrocephalus" represents a landmark recognition of a treatable dementia. This entity is characterized by spasticity with hyper-reflexia, ataxia, occasional urinary incontinence and dementia. Invariably, the spinal fluid pressure is normal, pneumoencephalography demonstrates dilated ventricles with failure of air to pass over the cerebral cortex, radioactive iodinated serum albumin (RISA) in-